GPC3 gene

glypican 3

Normal Function

The *GPC3* gene provides instructions for making a protein called glypican 3. This protein is one of several glypicans in humans, each of which consists of a core protein attached to long sugar molecules called heparan sulfate chains. Glypicans are anchored to the cell membrane, where they interact with a variety of other proteins outside the cell. Glypicans appear to play important roles in development before birth. These proteins are involved in numerous cell functions including regulating cell growth and division (cell proliferation), cell survival, cell movement (migration), and the process by which cells mature to carry out specific functions (differentiation).

Several studies have found that glypican 3 interacts with other proteins at the surface of cells to restrain cell proliferation. Researchers believe that in some cell types, glypican 3 may act as a tumor suppressor, which is a protein that prevents cells from growing and dividing in an uncontrolled way to form a cancerous tumor. Glypican 3 may also cause some types of cells to self-destruct (undergo apoptosis) when they are no longer needed, which can help keep growth in check.

Although glypican 3 is known primarily as an inhibitor of cell growth and cell division, in some tissues it appears to have the opposite effect. Research suggests that in certain types of cells, such as cells in the liver, glypican 3 may interact with proteins called growth factors to promote cell growth and cell division.

Health Conditions Related to Genetic Changes

Simpson-Golabi-Behmel syndrome

More than 40 mutations in the *GPC3* gene have been identified in people with Simpson-Golabi-Behmel syndrome. Some of these mutations delete part or all of the gene, which prevents cells from producing functional glypican 3. Other mutations insert or delete a small amount of genetic material in the *GPC3* gene, or change one or a few protein building blocks (amino acids) used to make glypican 3. These mutations change the structure of the protein.

Mutations in the *GPC3* gene prevent glypican 3 from performing its usual functions, which may contribute to an increased rate of cell proliferation starting before birth. It is unclear, however, how a shortage of functional glypican 3 leads to overgrowth of the entire body and the other abnormalities characteristic of Simpson-Golabi-Behmel syndrome.

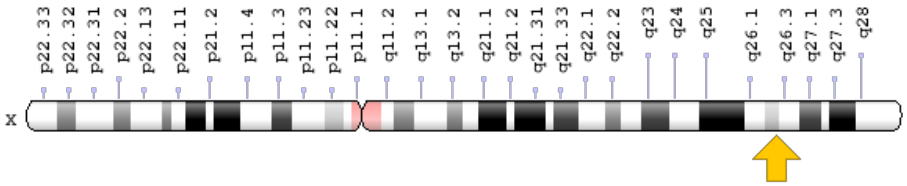
cancers

Changes in the activity (expression) of the *GPC3* gene have been associated with several forms of cancer. In particular, this gene is abnormally active (overexpressed) in a form of liver cancer called hepatocellular carcinoma. The increased gene expression may lead to uncontrolled cell growth and cell division in liver cells, which can result in the development of a cancerous tumor. On the other hand, a decrease in *GPC3* gene expression has been found in some ovarian cancers, breast cancers, colon cancers, and mesotheliomas (cancerous tumors that arise in the lining of the chest or abdomen).

Chromosomal Location

Cytogenetic Location: Xq26.2, which is the long (q) arm of the X chromosome at position 26.2

Molecular Location: base pairs 133,535,745 to 133,985,646 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DGSX
- glypican-3
- glypican proteoglycan 3
- GPC3_HUMAN
- GTR2-2
- Intestinal protein OCI-5
- MXR7
- OCI-5
- SDYS
- SGB

- SGBS
- SGBS1

Additional Information & Resources

Educational Resources

- National Cancer Institute: Liver Cancer Home Page
  https://www.cancer.gov/types/liver

GeneReviews

- Simpson-Golabi-Behmel Syndrome Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1219

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GPC3%5BTIAB%5D%29+OR+%28glypican+3%5BTIAB%5D%29%29+OR+%28glypican-3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- GLYPICAN 3
  http://omim.org/entry/300037
- HEPATOCELLULAR CARCINOMA
  http://omim.org/entry/114550

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GPC3ID156.html
- Cancer Genetics Web
  http://www.cancerindex.org/geneweb/GPC3.htm
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GPC3%5Bgene%5D
- HGNC Gene Family: Glypicans
  http://www.genenames.org/cgi-bin/genefamilies/set/572
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4451

- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2719
- UniProt
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